



## popliteal pterygium syndrome

Popliteal pterygium syndrome is a condition that affects the development of the face, skin, and genitals. Most people with this disorder are born with a cleft lip, a cleft palate (an opening in the roof of the mouth), or both. Affected individuals may have depressions (pits) near the center of the lower lip, which may appear moist due to the presence of salivary and mucous glands in the pits. Small mounds of tissue on the lower lip may also occur. In some cases, people with popliteal pterygium syndrome have missing teeth.

Individuals with popliteal pterygium syndrome may be born with webs of skin on the backs of the legs across the knee joint, which may impair mobility unless surgically removed. Affected individuals may also have webbing or fusion of the fingers or toes (syndactyly), characteristic triangular folds of skin over the nails of the large toes, or tissue connecting the upper and lower eyelids or the upper and lower jaws. They may have abnormal genitals, including unusually small external genital folds (hypoplasia of the labia majora) in females. Affected males may have undescended testes (cryptorchidism) or a scrotum divided into two lobes (bifid scrotum).

People with popliteal pterygium syndrome who have cleft lip and/or palate, like other individuals with these facial conditions, may have an increased risk of delayed language development, learning disabilities, or other mild cognitive problems. The average IQ of individuals with popliteal pterygium syndrome is not significantly different from that of the general population.

### Frequency

Popliteal pterygium syndrome is a rare condition, occurring in approximately 1 in 300,000 individuals.

### Genetic Changes

Mutations in the *IRF6* gene cause popliteal pterygium syndrome. The *IRF6* gene provides instructions for making a protein that plays an important role in early development. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes.

The IRF6 protein is active in cells that give rise to tissues in the head and face. It is also involved in the development of other parts of the body, including the skin and genitals.

Mutations in the *IRF6* gene that cause popliteal pterygium syndrome may change the transcription factor's effect on the activity of certain genes. This affects the development

and maturation of tissues in the face, skin, and genitals, resulting in the signs and symptoms of popliteal pterygium syndrome.

### **Inheritance Pattern**

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

### **Other Names for This Condition**

- Facio-genito-popliteal syndrome
- PPS

### **Diagnosis & Management**

#### Genetic Testing

- Genetic Testing Registry: Popliteal pterygium syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265259/>

#### Other Diagnosis and Management Resources

- GeneReview: IRF6-Related Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1407>

#### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

### **Additional Information & Resources**

#### MedlinePlus

- Health Topic: Cleft Lip and Palate  
<https://medlineplus.gov/cleftlipandpalate.html>

### Genetic and Rare Diseases Information Center

- Popliteal pterygium syndrome  
<https://rarediseases.info.nih.gov/diseases/3242/popliteal-ptyergium-syndrome>

### Educational Resources

- Disease InfoSearch: Popliteal pterygium syndrome  
<http://www.diseaseinfosearch.org/Popliteal+pterygium+syndrome/5870>
- MalaCards: popliteal pterygium syndrome 1  
[http://www.malacards.org/card/popliteal\\_pterygium\\_syndrome\\_1](http://www.malacards.org/card/popliteal_pterygium_syndrome_1)
- March of Dimes: Cleft Lip and Cleft Palate  
<http://www.marchofdimes.org/baby/cleft-lip-and-cleft-palate.aspx>

### Patient Support and Advocacy Resources

- AboutFace International  
<http://www.aboutface.ca/>
- Children's Craniofacial Association  
<http://www.ccakids.com/>
- Cleft Palate Foundation  
<http://www.cleftline.org/>

### GeneReviews

- IRF6-Related Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1407>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Abnormalities,+Multiple%5BMAJR%5D%29+AND+%28%28popliteal+pterygium+syndrome%5BTIAB%5D%29+OR+%28facio-genito-popliteal+syndrome%5BTIAB%5D%29+OR+%28pps%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- POPLITEAL PTERYGIUM SYNDROME  
<http://omim.org/entry/119500>

## Sources for This Summary

- GeneReview: IRF6-Related Disorders  
<https://www.ncbi.nlm.nih.gov/books/NBK1407>
  - Ghassibé M, Revencu N, Bayet B, Gillerot Y, Vanwijck R, Verellen-Dumoulin C, Vikkula M. Six families with van der Woude and/or popliteal pterygium syndrome: all with a mutation in the IRF6 gene. *J Med Genet*. 2004 Feb;41(2):e15.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14757865>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1735675/>
  - Kondo S, Schutte BC, Richardson RJ, Bjork BC, Knight AS, Watanabe Y, Howard E, de Lima RL, Daack-Hirsch S, Sander A, McDonald-McGinn DM, Zackai EH, Lammer EJ, Aylsworth AS, Ardinger HH, Lidral AC, Pober BR, Moreno L, Arcos-Burgos M, Valencia C, Houdayer C, Bahuau M, Moretti-Ferreira D, Richieri-Costa A, Dixon MJ, Murray JC. Mutations in IRF6 cause Van der Woude and popliteal pterygium syndromes. *Nat Genet*. 2002 Oct;32(2):285-9. Epub 2002 Sep 3.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12219090>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3169431/>
  - OMIM: POPLITEAL PTERYGIUM SYNDROME  
<http://omim.org/entry/119500>
- 

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/popliteal-ptyerygium-syndrome>

Reviewed: April 2008

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services